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Figure 6. The genomic sequence for *IKBKAP* (SEQ ID NO: 1).

Figure 7- The cDNA sequence for *IKBKAP* (SEQ ID NO: 2)

Figure 8- the amino acid sequence of the *IKBKAP* gene (SEQ ID NO: 3)

Figure 9- Comparison of the amino acid sequence of Ikap across several species (SEQ ID NOS 4-9, respectively, in order of appearance). Alignment of the amino acid sequence of Ikap (M_musculus) with that of *Homo sapiens* (H_sapiens), *Drosophila melanogaster* (D_melanogaster), *Saccharomyces cerevisiae* (S_cervisiae), *Arabidopsis thaliana* (A_thaliana), and *Caenorhabditis elegans* (C_elegans).

Figure 10- Comparison of the Novel Mouse *Ikbkap* Gene with Multiple Species Homologs

Figure 11- Mouse *Ikbkap* Exon and Intron Boundaries (Acceptor site sequences have been assigned SEQ ID NOS 10-45, respectively, in order of appearance. Donor site sequences have been assigned SEQ ID NOS 46-81, respectively, in order of appearance).

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Exon trapping experiments of cosmids from a physical map of the candidate region yielded 5 exons that were used to screen a human frontal cortex cDNA library. Several cDNA clones were isolated and assembled into a novel transcript encoding a 1332 AA protein that was later identified as *IKBKAP* (Cohen et al. 1998). The complete 5.9 kb cDNA sequence of *IKBKAP* has been submitted to GenBank under accession number AF153419. In order to screen for mutations in FD patients, total lymphoblast RNA was reverse transcribed and overlapping sections of *IKBKAP* were amplified by PCR and sequenced. Evaluation of the splicing defect was performed using the following primers: 18F: GCCAGTGTTTTTGCCTGAG (SEQ ID NO: 82); 19F: CGGATTGTCACTGTTGTGC (SEQ ID NO: 83); 23R: GACTGCTCTCATAGCATCGC (SEQ ID NO: 84) (Fig. 1).

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In a preferred embodiment, the amplification primers used for detecting the T-C mutation and the G-C mutation in the FD gene are 5'- GCCAGTGTTTTTGCCTGAG – 3' (SEQ ID NO: 82) / 5'-GACTGCTCTCATAGCATCGC- 3' (SEQ ID NO: 84) and 5'-CGGATTGTCACTGTTGTGC- 3' (SEQ ID NO: 83) / 5'-GACTGCTCTCATAGCATCGC-3, (SEQ ID NO: 84) respectively.

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Following PCR amplification, the PCR products are subjected to a hybridization assay using allele-specific oligonucleotides. In a preferred embodiment, the allele-specific oligonucleotides used to detect the mutatons in the FD gene are as follows:

5'- AAGTAAG(T/C)GCCATTG- 3' (SEQ ID NO: 85) and 5'-GGTTCAC(G/C)GATTGTC (SEQ ID NO: 86).